Health Care Provider Fact Sheet

Disease Name — Citrullinemia

Alternate name(s)	Argininosuccinic acid synthetase deficiency
Acronym	ASAS
Disease Classification	Amino Acid Disorder
Variants	Yes
Variant name	Citrullinemia type II (adult and neonatal onset forms) – caused by *SLC25A13* mutations
Symptom onset	Neonatal with some variability
Symptoms	Potential lethal coma, seizures, anorexia, vomiting, lethargy, apnea and hypertonia. Possible enlarged liver.
Natural history without treatment	Mental retardation due to hyperammonemia.
Natural history with treatment	Normal IQ and development are possible if no damage from initial or subsequent hyperammonemic episodes.
Treatment	Management of hyperammonemic cases with sodium benzoate and/or phenylacetate and arginine. Dietary restriction of protein, arginine and essential amino acid supplementation.
Emergency Medical Treatment	See sheet from American College of Medical Genetics (attached) or for more information, go to website: http://www.acmg.net/StaticContent/ACT/Citrullinemia.pdf
Physical phenotype	None
Inheritance	Autosomal recessive
General population incidence	Rare
Ethnic differences	Yes
Population	Citrullinemia type II is common in Japan
Ethnic incidence	N/A
Enzyme location	Widely expressed in tissues; liver, kidney and fibroblasts.
Enzyme Function	Catalyzes the conversion of citrulline and aspartic acid to argininosuccinic acid.
Missing Enzyme	Argininosuccinic acid synthetase
Metabolite changes	Hyperammonemia
Prenatal testing	Linkage analysis and enzyme testing
MS/MS Profile	N/A
OMIM Link	http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=215700
Genetests Link	www.genetests.org
Support Group	National Urea Cycle Disorders Foundation http://www.nucdf.org/ National Coalition for PKU and Allied Disorders http://www.pku-allieddisorders.org/ Children Living with Inherited Metabolic Diseases http://www.climb.org.uk/

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